



## IRF6 gene

interferon regulatory factor 6

### Normal Function

The *IRF6* gene provides instructions for making a protein that plays an important role in early development. This protein is a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of particular genes.

The IRF6 protein is active in cells that give rise to tissues in the head and face. It is also involved in the development of other parts of the body, including the skin and genitals.

### Health Conditions Related to Genetic Changes

#### popliteal pterygium syndrome

Mutations in the *IRF6* gene that cause popliteal pterygium syndrome may change the transcription factor's effects on the activity of certain genes. This affects the development and maturation of tissues in the face, skin, and genitals, resulting in the facial and genital abnormalities, skin webbing, and fusion of the fingers or toes (syndactyly) seen in popliteal pterygium syndrome.

#### van der Woude syndrome

Mutations in the *IRF6* gene that cause van der Woude syndrome prevent one copy of the gene in each cell from making any functional protein. A shortage of the IRF6 protein affects the development and maturation of tissues in the skull and face. These abnormalities underlie the signs and symptoms of van der Woude syndrome, including cleft lip, cleft palate (an opening in the roof of the mouth), and pits or mounds in the lower lip.

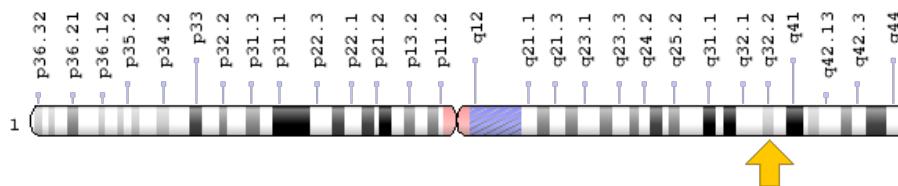
#### other disorders

Certain variations in the *IRF6* gene have been associated with increased risk of cleft lip, cleft palate, or both. When these features appear without other signs or symptoms, the condition is called isolated cleft lip and/or palate. The *IRF6* gene variations are believed to affect the function of the IRF6 protein in its role as a transcription factor, which may interfere with the normal development of the face.

## Chromosomal Location

Cytogenetic Location: 1q32.2, which is the long (q) arm of chromosome 1 at position 32.2

Molecular Location: base pairs 209,785,623 to 209,806,175 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- IRF6\_HUMAN
- LPS
- OFC6
- PIT
- PPS
- VWS
- VWS1

## Additional Information & Resources

### GeneReviews

- IRF6-Related Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK1407>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28IRF6%5BTIAB%5D%29+OR+%28interferon+regulatory+factor+6%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

## OMIM

- INTERFERON REGULATORY FACTOR 6  
<http://omim.org/entry/607199>
- OROFACIAL CLEFT 6, SUSCEPTIBILITY TO  
<http://omim.org/entry/608864>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_IRF6.html](http://atlasgeneticsoncology.org/Genes/GC_IRF6.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=IRF6%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=6121](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6121)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/3664>
- UniProt  
<http://www.uniprot.org/uniprot/O14896>

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